Testimony before the Public Witness Hearing, March 18, 2009
House Appropriations Subcommittee on
Labor-Health and Human Services-Education
by
Neil Horikoshi, Chairman of the Board
Aplastic Anemia & MDS International Foundation

Thank you, Mr. Chairman and members of the subcommittee, for giving me the opportunity to testify today. Many of you have probably never heard of aplastic anemia or some of the other diseases that are collectively known as "bone marrow failure diseases." I certainly had never heard of these diseases prior to February 29, 2000. That day, however, would prove fateful for me and change my life in a way I could not have foreseen. And over a critical 48 hour stretch, I would descend from a level of seemingly perfect health to the precipice of death. I would soon learn firsthand how a rare, deadly bone marrow failure disease can strike anyone, regardless of age or state of general health.

In 2000, I was on international assignment with IBM in Japan, and I traveled quite frequently for this job. Prior to a scheduled trip to India, I was feeling exceptionally fatigued. I also started to suffer from a shortness of breath, constant chills, and tingling in his fingers and toes. I even noticed red splotches on both of my calves.

These symptoms should have set off sirens and whistles in any person. Yet, as a healthy 40+ year old male who exercised and ate well, I shrugged it off. "Maybe I should get more exercise, or more sleep, or take some vitamins," I recall saying to myself at the time.

If I had not had a regularly scheduled physical examination prior to this trip -- the last available Saturday appointment at a clinic in Tokyo – I may never have made it alive to India. My fortuitous decision to keep my appointment led to a series of steps that would determine my ultimate diagnosis and the treatment that would save my life.

At my physical, the medical doctor ordered a Complete Blood Count (CBC) immediately, and noticed all the telltale symptoms of a serious medical condition. The next day --- the day before my scheduled trip -- I experienced worsening symptoms. I began to have blurred vision in my left eye due to a small hemorrhage behind my retina. My doctor called me that day and clearly stated: "cancel your trip to India….your blood counts are unusually low." Fortunately, I listened to the advice of my doctor.

The blood specialist at the local hospital in Japan conducted another CBC, and he reconfirmed that I had a serious blood shortage. His best guess diagnosis was that I might have leukemia. I was given a blood transfusion, sufficient to put me on a flight to Honolulu for further treatment.

In a classic case of "good news, bad news," the hematologist in Hawaii informed that I did not have leukemia. The bad news was that I was diagnosed with a rare bone marrow failure disease known as **aplastic anemia**, which in laymen's terms meant that my bone marrow was not producing any blood.

I had no idea what this diagnosis meant, what the treatment was, or how I contracted the disease. As I would later discover, the causes of aplastic anemia and other bone marrow failure diseases are unknown. Bone marrow failure disease has been linked to environmental factors to which we are all commonly exposed. Researchers suspect that undefined genetic factors make some individuals more susceptible to bone marrow disease.

My treatment in the short term consisted of getting ongoing CBC's to monitor my blood counts. As the red and platelet blood counts dropped into a trough range, I required ongoing blood transfusions of both red and platelet blood. (Red blood cells support the flow of oxygen in one's body; platelets support positive healing of all wounds and bleeding; white blood cells are required to fight infections, and one's body must be able to produce these cells.)

Long term options were somewhat less clear.

I was advised that I was too old for a bone marrow transplant. A 40+ year old patient requires the anti-thymocyte globulin (ATG) treatment. My family doctor told me it may be wise to 'get your things in order.' Those were five words that shaped my choice in the weeks and months ahead."

Like many Americans who are diagnosed with a rare disease, I turned to the Internet for information about treatment options and support networks. I discovered a unique resource and positive voice of reinforcement in the Aplastic Anemia & MDS International Foundation. The organization provided me with an abundance of patient-friendly information and a network of medical doctors who could provide answers to basic and personal questions about the treatment I faced.

The Foundation also directed me to Dr. Neal Young, the pre-eminent expert on aplastic anemia at the National Institutes of Health (NIH). Dr. Young pioneered research in this orphan disease, and his success with immunosuppressant treatments literally saved my life and the lives of many others.

By the end of March, 2000, within a month of my initial diagnosis, I was treated with ATG in a local Honolulu hospital. I was not transfusion free until May, 2000 when my blood counts stabilized and I slowly began to produce blood. I returned to my job in August 2000 and felt I was a very lucky person. In January 2001, I showed signs of relapsing and got a second ATG treatment in Hawaii, and within a week, my blood counts shot up to the normal levels they are at today.

What I learned from my medical consultations with Dr. Young, and my interaction with the Foundation, was that I had hope. Not more than a decade ago, most people diagnosed with acute aplastic anemia had little chance of survival. Today the success and survival rate is viewed positively, thanks in large part to the research funded by NIH

While many advances have been made in the understanding and treatment of bone marrow failure diseases, we still do not have a cure. Every year, more than 20,000 Americans are diagnosed with aplastic anemia, myelodysplastic syndromes or PNH – the three primary bone marrow failure diseases. We still do not have a good handle on what causes these diseases, and why some groups, such as ethnic Asians, are affected more than others.

My life was saved thanks to medical research – and today I want to use my story to help save the lives of others who are affected by this terrible disease. Today, I serve as the Chairman of the Board of the Foundation, and am committed to advocating for greater research and awareness that will ultimately lead to a cure.

I therefore urge the Labor-HHS-Education Subcommittee to commit greater resources to NIH and the Centers for Disease Control and Prevention. I also urge every member of this committee to support H.R. 1230, the Bone Marrow Failure Disease Research and Treatment Act, to increase surveillance efforts at CDC so that we can truly understand what is causing these diseases. This legislation has been introduced by Representative Doris Matsui (D-CA), who only a few years ago lost her husband – and your former colleague – Bob Matsui to MDS, one of the bone marrow failure diseases. When this legislation is enacted, I hope this committee will provide the necessary appropriations to implement better surveillance at the CDC.

Finally, I would also like to recognize the full committee for its support of bone marrow failure disease research at the Department of Defense (DoD). Although not within the jurisdiction of this subcommittee, the DoD Bone Marrow Failure Disease Research program was funded at $5 million in fiscal year 2009. This funding will have a great impact on gaining a better understanding of these diseases, particularly within the veteran population.

Mr. Chairman, I am a very lucky person. Because of early intervention and access to critical information and support groups, I survived this awful disease. I hope that my testimony today can in some small way make a difference for everyone suffering from bone marrow failure diseases. I urge the House Appropriations Committee to continue its support for bone marrow failure disease research and surveillance at NIH, CDC and DoD.